

Amyloidosis

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In news— Former Pakistan President Pervez Musharraf (retired) has been affected by Amyloidosis recently.

What is Amyloidosis?

- Amyloidosis is a **rare disease that occurs when an abnormal protein, called amyloid, builds up in one's organs, affecting their shape and functioning.**
- **Amyloid deposits can build up in the heart, brain, kidneys, spleen** and other parts of the body, leading to life-threatening conditions like **organ failure**.
- **Amyloid isn't normally found in the body, but can be formed from several different types of proteins.** Some varieties of amyloidosis occur in association with other diseases.
- These types may improve with treatment of the underlying disease. Some varieties of amyloidosis may lead to life-threatening organ failure.
- **Amyloidosis may be secondary to a different health condition** or can develop as a primary condition as well.
- Sometimes, **it is due to a mutation in a gene**, but usually, the cause of amyloidosis remains unknown.
- **Many different proteins can lead to amyloid deposits**, but only a few lead to major health problems.
- **The type of protein and where it collects tell the type of amyloidosis one has.** Amyloid deposits may collect throughout one's body or in just one area.
- Moreover, **while some varieties are hereditary, others are caused by outside factors**, such as inflammatory diseases or long-term dialysis.
- **Symptoms of amyloidosis are often subtle and can vary greatly** depending on where the amyloid protein is collecting in the body.
- **As amyloidosis progresses, the deposits of amyloid can**

harm the heart, liver, spleen, kidneys, digestive tract, brain or nerves.

The general signs or symptoms would include:

- Severe fatigue.
- Loss of weight.
- Swelling in the belly, legs, ankles or feet.
- Numbness, pain or tingling in hands or feet.
- Changes in skin colour.
- Purple spots (purpura) or bruised-looking areas of skin around the eyes.
- Bleeding more than usual after an injury.
- Swelling of the tongue.
- Shortness of breath.

There are also different amyloidosis that are prevalent:

Light-chain (AL) amyloidosis-

- It is the **most common type in developed countries** which can affect the kidneys, spleen, heart, and other organs.
- People with conditions such as multiple myeloma or a bone marrow illness are more likely to have AL amyloidosis.
- This **starts in plasma cells within the bone marrow**. Plasma cells create antibodies with both heavy chain and light chain proteins.
- If the plasma cells undergo abnormal changes, they produce excess light chain proteins that can end up in the bloodstream.
- These damaged protein bits can accumulate in the body's tissues and damage vital organs such as the heart.

AA amyloidosis-

- **Previously known as secondary amyloidosis, this condition is the result of another chronic infectious or**

inflammatory disease, such as **rheumatoid arthritis**, Crohn's disease, or ulcerative colitis. It mostly affects one's kidneys, digestive tract, liver, and heart.

- AA refers to the amyloid type A protein that causes it.

Dialysis-related amyloidosis-

- It is **more common in older adults** and people who have been on dialysis for more than 5 years.
- This form of amyloidosis is **caused by deposits of beta-2 microglobulin** that build up in the blood.
- Deposits can build up in many different tissues, but it most commonly affects bones, joints, and tendons.

Transthyretin amyloidosis-

- It **can be inherited from a family member and is hence commonly referred to as familial amyloidosis**. Transthyretin is a protein that is also known as prealbumin made in the liver.
- As such, this often affects the liver, nerves, heart, and kidneys and many genetic defects are linked to a higher chance of amyloid disease.